

Contents of Volume 58

January 1996

1995 WILLIAM ALLAN AWARD ADDRESS

- 1 Human Genetics: A Discipline at Risk for Fragmentation**
Kurt Hirschhorn

Invited Editorials

- 7 Absence Makes the Search Grow Longer**
W. B. Dobyns
- 17 Nutritional Ecogenetics: Homocysteine-Related Arteriosclerotic Vascular Disease, Neural Tube Defects, and Folic Acid**
Arno G. Motulsky

Review

- 21 Chromosomal Duplications in Bacteria, Fruit Flies, and Humans**
James R. Lupski, John R. Roth, and George M. Weinstock

Original Articles

- 28 The Gene Responsible for a Severe Form of Peripheral Neuropathy and Agenesis of the Corpus Callosum Maps to Chromosome 15q**
Leanne K. Casaubon, Michel Melanson, Iscia Lopes-Cendes, Claude Marineau, Eva Andermann, Frederick Andermann, Jean Weissenbach, Claude Prévost, Jean-Pierre Bouchard, Jean Mathieu, and Guy A. Rouleau
- 35 Molecular Genetic Analysis in Mild Hyperhomocysteinemia: A Common Mutation in the Methylenetetrahydrofolate Reductase Gene Is a Genetic Risk Factor for Cardiovascular Disease**
Leo A. J. Kluijtmans, Lambert P. W. J. van den Heuvel, Godfried H. J. Boers, Phyllis Frosst, Erik M. B. Stevens, Bernard A. van Oost, Martin den Heijer, Frans J. M. Trijbels, Rima Rozen, and Henk J. Blom
- 42 A High Incidence of BRCA1 Mutations in 20 Breast-Ovarian Cancer Families**
Olga Serova, Marco Montagna, Delphine Torchard, Steven A. Narod, Patricia Tonin, Bakary Sylla, Henry T. Lynch, Jean Feunteun, and Gilbert M. Lenoir
- 52 Characterization of 12 Silent Alleles of the Human Butyrylcholinesterase (BCHE) Gene**
S. L. Primo-Parmo, C. F. Bartels, B. Wiersema, A. F. L. van der Spek, J. W. Innis, and B. N. La Du
- 65 Analysis of a Splice-Site Mutation in the *sap*-Precursor Gene of a Patient with Metachromatic Leukodystrophy**
M. Henseler, A. Klein, M. Reber, M. T. Vanier, P. Landrieu, and K. Sandhoff
- 75 Molecular Genetic Defect Underlying α -L-Iduronidase Pseudodeficiency**
Elena L. Aronovich, Dao Pan, and Chester B. Whitley
- 86 Screening the 3' Region of the Polycystic Kidney Disease 1 (PKD1) Gene Reveals Six Novel Mutations**
Belén Peral, José L. San Millán, Albert C. M. Ong, Vicki Gamble, Christopher J. Ward, Carol Strong, and Peter C. Harris

- 97 Mutation Analysis of Very-Long-Chain Acyl-Coenzyme A Dehydrogenase (VLCAD) Deficiency: Identification and Characterization of Mutant VLCAD cDNAs from Four Patients**
Masayoshi Souri, Toshifumi Aoyama, Kohji Oriti, Seiji Yamaguchi, and Takashi Hashimoto
- 107 Identification of 13 New Mutations in the Vasopressin-Neurophysin II Gene in 17 Kindreds with Familial Autosomal Dominant Neurohypophyseal Diabetes Insipidus**
Søren Rittig, Gary L. Robertson, Charlotte Siggaard, László Kovács, Niels Gregersen, Jens Nyborg, and Erling B. Pedersen
- 118 Genetic Heterogeneity in Niemann-Pick C Disease: A Study Using Somatic Cell Hybridization and Linkage Analysis**
M. T. Vanier, S. Duthel, C. Rodriguez-Lafrasse, P. Pentchev, and E. D. Carstea
- 126 Fine Mapping of the EDA Gene: A Translocation Breakpoint Is Associated with a CpG Island That Is Transcribed**
Anand K. Srivastava, Outi Montonen, Ulpu Saarialho-Kere, Ellson Chen, Primo Baybayan, Johanna Pispas, Janusz Limon, David Schlessinger, and Juha Kere
- 133 Molecular Definition of Red Cell Rh Haplotypes by Tightly Linked *SphI* RFLPs**
C.-H. Huang, M. E. Reid, Y. Chen, G. Coghlan, and Y. Okubo
- 143 *PRB1*, *PRB2*, and *PRB4* Coded Polymorphisms among Human Salivary Concanavalin-A Binding, II-1, and Po Proline-Rich Proteins**
E. A. Azen, E. Amberger, S. Fisher, A. Prakobphol, and R. L. Niece
- 154 Molecular Definition of Breakpoints Associated with Human Xq Isochromosomes: Implications for Mechanisms of Formation**
Dayna J. Wolff, Andrew P. Miller, Daniel L. Van Dyke, Stuart Schwartz, and Huntington F. Willard
- 161 Lack of X Inactivation Associated with Maternal X Isodisomy: Evidence for a Counting Mechanism Prior to X Inactivation during Human Embryogenesis**
Barbara R. Migeon, Peter Jeppesen, Beth S. Torchia, Sidong Fu, Melanie A. Dunn, Joyce Axelman, Barbara J. Schmeckpeper, Judy Fantes, Robert T. Zori, and Daniel J. Driscoll
- 171 Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy, Genetic Homogeneity, and Mapping of the Locus within a 2-cM Interval**
A. Ducros, T. Nagy, S. Alamowitch, A. Nibbio, A. Joutel, K. Vahedi, H. Chabriat, M. T. Iba-Zizen, J. Julien, P. Davous, J. Y. Goas, O. Lyon-Caen, B. Dubois, X. Ducrocq, F. Salsa, M. Ragno, P. Burkhard, C. Bassetti, M. Hutchinson, M. Verin, F. Viader, F. Chapon, M. Levasseur, J. L. Mas, O. Delrieu, J. Maciazek, M. Prieur, H. Mohrenweiser, J. F. Bach, M. G. Bousser, and E. Tournier-Lasserre
- 182 Familial Recurrence-Pattern Analysis of Nonsyndromic Isolated Cleft Palate—A Danish Registry Study**
Kaare Christensen and Laura E. Mitchell
- 191 Influence of Apolipoprotein E Genotype on the Transmission of Alzheimer Disease in a Community-Based Sample**
Gail Pairitz Jarvik, Eric B. Larson, Katrina Goddard, Walter A. Kukull, Gerard D. Schellenberg, and Ellen M. Wijsman
- 201 Statistical Models for Trisomic Phenotypes**
Neil E. Lamb, Eleanor Feingold, and Stephanie L. Sherman
- 213 Segregation Analysis of Continuous Phenotypes by Using Higher Sample Moments**
Hang Lee and Daniel O. Stram

225 Error Detection for Genetic Data, Using Likelihood Methods

Margaret Gelder Ehm, Marek Kimmel, and Robert W. Cottingham, Jr.

Obituary**235 Anita Harding (1952-95): In Memoriam**

J. Poulton and S. M. Huson

Letters to the Editor**237 Reverse Mutation in Fragile X Syndrome**

Guillermo Antiñolo, Salud Borrego, Juan C. Cabeza, Rosario Sánchez, Javier Sánchez, and Beatriz Sánchez

239 Exclusion of Linkage between Cleft Lip With or Without Cleft Palate and Markers on Chromosomes 4 and 6

Susan Halloran Blanton, Eric Crowder, Sue Malcolm, Robin Winter, David L. Gasser, Samuel Stal, John Mulliken, and Jacqueline T. Hecht

241 Multiple Mutations in a Specific Gene in a Small Geographic Area: A Common Phenomenon?

Joël Zlotogora, Volkmar Gieselmann, and Gideon Bach

243 The Distinction between Juvenile and Adult-Onset Primary Open-Angle Glaucoma

Janey L. Wiggs, Karim F. Damji, Jonathan L. Haines, Margaret A. Pericak-Vance, and R. Rand Allingham

Book Reviews**245 Reproductive Effects of Chemical, Physical, and Biologic Agents: REPROTOX[®].** By Anthony R. Scialli,

Armand Lione, and G. K. Boyle Padgett

Reviewed by J. M. Friedman

245 Pregnancy in a High-Tech Age: Paradoxes of Choice. By Robin Gregg

Reviewed by Beth A. Fine

247 The Golden Helix: Inside Biotech Ventures. By Arthur Kornberg

Reviewed by Richard Gelinas

Announcements**249 Employment and Fellowship Opportunities; International Congress; Course; Call for Abstracts; DNA Standards; Familial Breast Cancer Samples; Call for Patients****252 Errata****Information for Contributors**

February 1996

Original Articles**255 Atelosteogenesis Type II Is Caused by Mutations in the Diastrophic Dysplasia Sulfate-Transporter Gene (DTDST): Evidence for a Phenotypic Series Involving Three Chondrodysplasias**

Johanna Hästbacka, Andrea Superti-Furga, William R. Wilcox, David L. Rimoin, Daniel H. Cohn, and Eric S. Lander

- 263 Defects in the DNA Repair and Transcription Gene *ERCC2(XPD)* in Trichothiodystrophy**
Kyoko Takayama, Edmund P. Salazar, Bernard C. Broughton, Alan R. Lehmann, Alain Sarasin, Larry H. Thompson, and Christine A. Weber
- 271 Haplotype and Phenotype Analysis of Six Recurrent *BRCA1* Mutations in 61 Families: Results of an International Study**
Susan L. Neuhausen, Sylvie Mazoyer, Lori Friedman, Michael Stratton, Ken Offit, Adelaide Caligo, Gail Tomlinson, Lisa Cannon-Albright, Tim Bishop, David Kelsell, Ellen Solomon, Barbara Weber, Fergus Couch, Jeffery Struewing, Patricia Tonin, Francine Durocher, Steven Narod, Mark H. Skolnick, Gilbert Lenoir, Olga Serova, Bruce Ponder, Dominique Stoppa-Lyonnet, Douglas Easton, Mary-Claire King, and David E. Goldgar
- 281 A 10-bp Deletion in the Apolipoprotein ϵ Gene Causing Apolipoprotein E Deficiency and Severe Type III Hyperlipoproteinemia**
Giso Feussner, Jürgen Dobmeyer, Hermann-Josef Gröne, Stefan Lohmer, and Stefan Wohlfeil
- 292 Modulation of the Phenotype in Dominant Erythropoietic Protoporphria by a Low Expression of the Normal Ferrochelatase Allele**
L. Gouya, J. Ch. Deybach, J. Lamoril, V. Da Silva, C. Beaumont, B. Grandchamp, and Y. Nordmann
- 300 Majority of *hMLH1* Mutations Responsible for Hereditary Nonpolyposis Colorectal Cancer Cluster at the Exonic Region 15-16**
Juul Wijnen, P. Meera Khan, Hans Vasen, Fred Menko, Heleen van der Klift, Marianne van den Broek, Inge van Leeuwen-Cornelisse, Fokko Nagengast, E. J. Meijers-Heijboer, Dick Lindhout, Gerrit Griffioen, Annemieke Cats, Jan Kleibeuker, Liliana Varesco, Lucio Bertario, Marie-Luise Bisgaard, Jan Mohr, Richard Kolodner, and Riccardo Fodde
- 308 Molecular Analysis of a Series of Alleles in Humans with Reduced Activity at the Triosephosphate Isomerase Locus**
M. Watanabe, B. C. Zingg, and H. W. Mohrenweiser
- 317 Novel Muscle Chloride Channel Mutations and Their Effects on Heterozygous Carriers**
Volker Mailänder, Roland Heine, Feza Deymeer, and Frank Lehmann-Horn
- 325 Longitudinal Study of a Heteroplasmic 3460 Leber Hereditary Optic Neuropathy Family by Multiplexed Primer-Extension Analysis and Nucleotide Sequencing**
S. S. Ghosh, E. Fahy, I. Bodis-Wollner, J. Sherman, and Neil Howell
- 335 Gene Structure, DNA Methylation, and Imprinted Expression of the Human *SNRPN* Gene**
Christopher C. Glenn, Shinji Saitoh, Michelle T. C. Jong, Michelle M. Filbrandt, Urvashi Surti, Daniel J. Driscoll, and Robert D. Nicholls
- 347 Map Refinement of Locus *RP13* to Human Chromosome 17p13.3 in a Second Family with Autosomal Dominant Retinitis Pigmentosa**
Tracy L. Kojis, Camilla Heinzmann, Pamela Flodman, Julielani T. Ngo, Robert S. Sparkes, M. Anne Spence, J. Bronwyn Bateman, and John R. Heckenlively
- 356 Aneuploidy in Human Sperm: The Use of Multicolor FISH to Test Various Theories of Nondisjunction**
E. L. Spriggs, A. W. Rademaker, and R. H. Martin
- 363 Genetic Evidence for the Neuronal Nitric Oxide Synthase Gene (*NOS1*) as a Susceptibility Locus for Infantile Pyloric Stenosis**
Eddie Chung, David Curtis, Gong Chen, Philip A. Marsden, Rebecca Twells, Weiming Xu, and Mark Gardiner
- 371 Sex Influences on the Penetrance of HLA Shared-Epitope Genotypes for Rheumatoid Arthritis**
Joanne M. Meyer, Jinfeng Han, Rovinder Singh, and George Moxley

384 Major-Locus Contributions to Variability of the Craniofacial Feature Dystopia Canthorum in Waardenburg Syndrome

Jennifer E. Reynolds, Mary L. Marazita, Joanne M. Meyer, Cathy A. Stevens, Lindon J. Eaves, Kathleen S. Amos, Lynn M. Ploughman, Charles MacLean, Walter E. Nance, and Scott R. Diehl

393 The Need for Anonymous Genetic Counseling and Testing

Maxwell J. Mehlman, Eric D. Kodish, Peter Whitehouse, Arthur B. Zinn, Sharmon Sollitto, Joshua Berger, Emmeline J. Chiao, Melissa S. Dosick, and Suzanne B. Cassidy

398 Establishing the Robustness of Short-Tandem-Repeat Statistics for Forensic Applications

Ian W. Evett, Peter D. Gill, John K. Scrannage, and B. S. Weir

408 Gametogenesis Processes and Multilocus Gene Identity by Descent

Sun-Wei Guo

Letters to the Editor

420 A Novel Mutation (Gln266→His) in the Alpha1 Subunit of the Inhibitory Glycine-Receptor Gene (GLRA1) in Hereditary Hyperekplexia

Nicoletta Milani, Leda Dalprá, Alberto del Prete, Roberto Zanini, and Lidia Larizza

422 Alterations of Chromosome 11q13 in Cervical Carcinoma Cell Lines

Nicholas C. Popescu and Drazen B. Zimonjic

424 Reply to Popescu and Zimonjic

Rachel A. Jesudasan, M. Razaur Rahman, Glen A. Evans, and Eri S. Srivatsan

425 Association between Prostate Cancer in Black Americans and an Allele of the PADPRP Pseudogene Locus on Chromosome 13

Jennifer A. Doll, Brian K. Suarez, and Helen Donis-Keller

428 Multiple Independent Origins of the COII/tRNA^{Leu} Intergenic 9-bp mtDNA Deletion in Aboriginal Australians

David J. Betty, Amy N. Chin-Atkins, Lynn Croft, Michaela Sraml, and Simon Easta

433 Nomenclature for Inherited Diseases of the Retina

Chris F. Inglehearn and Alison J. Hardcastle

435 Reply to Inglehearn and Hardcastle: The Map Is Not the Territory

Stephen P. Daiger, Rachel E. McGuire, and John R. Heckenlively

436 Certificates of Confidentiality

J. Alexander Lowden

437 Reply to Lowden

Charles L. Earley and Louise C. Strong

Announcements

439 Employment Opportunities; Conference

Information for Contributors

March 1996

Original Articles

- 441 Founding *BRCA1* Mutations in Hereditary Breast and Ovarian Cancer in Southern Sweden**
O. Johannsson, E. A. Ostermeyer, S. Håkansson, L. S. Friedman, U. Johansson, G. Sellberg, K. Brøndum-Nielsen, V. Sele, H. Olsson, M.-C. King, and Å. Borg
- 451 Rapid Detection of Regionally Clustered Germ-Line *BRCA1* Mutations by Multiplex Heteroduplex Analysis**
Simon A. Gayther, Patricia Harrington, Paul Russell, Galina Kharkevich, R. F. Garkavtseva, Bruce A. J. Ponder, and the UKCCCR Familial Ovarian Cancer Study Group
- 457 Molecular Analysis of Patients with β -Glucuronidase Deficiency Presenting as Hydrops Fetalis or as Early Mucopolysaccharidosis VII**
Raf Vervoort, M. Rafiqul Islam, William S. Sly, Marie-Thérèse Zabet, Wim J. Kleijer, Amparo Chabas, Anthony Fensom, Elisabeth P. Young, Inge Liebaers, and Willy Lissens
- 472 Prevalence and Origin of De Novo Duplications in Charcot-Marie-Tooth Disease Type 1A: First Report of a De Novo Duplication with a Maternal Origin**
Ian P. Blair, Janet Nash, Melissa J. Gordon, and Garth A. Nicholson
- 477 Discordant Phenotype in Siblings with X-Linked Agammaglobulinemia**
Michael J. Bykowski, Robert N. Haire, Yuko Ohta, Huayang Tang, Sun-Sang J. Sung, Ekaterina S. Veksler, Jeffrey M. Greene, Shu Man Fu, Gary W. Litman, and Kathleen E. Sullivan
- 484 Somatic Mosaicism in a Patient with Neurofibromatosis Type 1**
Steven D. Colman, Sonja A. Rasmussen, Vu T. Ho, Corinne R. Abernathy, and Margaret R. Wallace
- 491 FGFR2 Exon IIIa and IIIc Mutations in Crouzon, Jackson-Weiss, and Pfeiffer Syndromes: Evidence for Missense Changes, Insertions, and a Deletion Due to Alternative RNA Splicing**
Gregory A. Meyers, Donald Day, Rosalie Goldberg, Donna L. Daentl, Kelly A. Przylepa, Liane J. Abrams, John M. Graham, Jr., Murray Feingold, John B. Moeschler, Eileen Rawnsley, Alan F. Scott, and Ethylin Wang Jabs
- 499 Splicing Mutation in the ATR-X Gene Can Lead to a Dysmorphic Mental Retardation Phenotype without α -Thalassemia**
Laurent Villard, Annick Toutain, Anne-Marie Lossi, Jozef Gecz, Claude Houdayer, Claude Moraine, and Michel Fontès
- 506 The Age of Human Mutation: Genealogical and Linkage Disequilibrium Analysis of the CLN5 Mutation in the Finnish Population**
T. Varilo, M. Savukoski, R. Norio, P. Santavuori, L. Peltonen, and I. Järvelä
- 513 *FMR1* in Global Populations**
Catherine B. Kunst, Chris Zerylnick, Laurie Karickhoff, Evan Eichler, Jennifer Bullard, Maryse Chalifoux, Jeanette J. A. Holden, Antonio Torroni, David L. Nelson, and Stephen T. Warren
- 523 Linkage Disequilibrium Mapping Places the Gene Causing Familial Mediterranean Fever Close to *D16S246***
Ernesto N. Levy, Yang Shen, Annie Kupelian, Leonid Kruglyak, Ivona Aksentijevich, Elon Pras, James E. Balow, Jr., Brett Linzer, Xiaoguang Chen, David A. Shelton, Deborah Gumucio, Mordechai Pras, Mordechai Shohat, Jerome I. Rotter, Nathan Fischel-Ghodsian, Robert I. Richards, and Daniel L. Kastner

- 535 High-Resolution Mapping of the Gene for Cystinosis, Using Combined Biochemical and Linkage Analysis**
Geneviève Jean, Arno Fuchshuber, Margaret M. Town, Olivier Gribouval, Jerry A. Schneider, Michel Broyer, William van't Hoff, Patrick Niaudet, and Corinne Antignac

- 544 Isolation of a Cosmid Clone Corresponding to an inv(21) Breakpoint of a Patient with Transient Abnormal Myelopoiesis**

Tohru Ohta, Motoi Nakano, Takahiro Tsujita, Kyohko Abe, Kazutoyo Osoegawa, Tetsushi Yamagata, Koh-ichiro Yoshiura, Yoshihiro Jinno, Eiichi Soeda, Yusuke Nakamura, and Norio Niikawa

- 551 Orofacial Clefts, Parental Cigarette Smoking, and Transforming Growth Factor-Alpha Gene Variants**

Gary M. Shaw, Cathy R. Wasserman, Edward J. Lammer, Cynthia D. O'Malley, Jeffrey C. Murray, Ann M. Basart, and Marie M. Tolarova

- 562 Heritability of Human Brain Functioning as Assessed by Electroencephalography**

C. E. M. van Beijsterveldt, P. C. M. Molenaar, E. J. C. de Geus, and D. I. Boomsma

- 574 Relative Risk of Alzheimer Disease and Age-at-Onset Distributions, Based on APOE Genotypes among Elderly African Americans, Caucasians, and Hispanics in New York City**

Ming-Xin Tang, Gladys Maestre, Wei-Yann Tsai, Xin-Hua Liu, Lin Feng, Wai-Yee Chung, Michael Chun, Peter Schofield, Yaakov Stern, Benjamin Tycko, and Richard Mayeux

- 585 Multilocus Genetic Determinants of LDL Particle Size in Coronary Artery Disease Families**

Jerome I. Rotter, Xiangdong Bu, Rita M. Cantor, Craig H. Warden, Jane Brown, Richard J. Gray, Patricia J. Blanche, Ronald M. Krauss, and Aldons J. Lusis

- 595 mtDNA Control-Region Sequence Variation Suggests Multiple Independent Origins of an "Asian-Specific" 9-bp Deletion in Sub-Saharan Africans**

Himla Soodyall, Linda Vigilant, Adrian V. Hill, Mark Stoneking, and Trefor Jenkins

- 609 Mitochondrial D-loop "Signatures" Produced by Low-Stringency Single Specific Primer PCR Constitute a Simple Comparative Human Identity Test**

Guillermo Barreto, Annamaria R. Vago, Charles Ginther, Andrew J. G. Simpson, and Sérgio D. J. Pena

- 617 Lack of Interest by Nonpregnant Couples in Population-Based Cystic Fibrosis Carrier Screening**

Ellen Wright Clayton, Vickie L. Hannig, Jean P. Pfothauer, Robert A. Parker, Preston W. Campbell III, and John A. Phillips III

- 628 Advanced Maternal Age and the Risk of Down Syndrome Characterized by the Meiotic Stage of the Chromosomal Error: A Population-Based Study**

Paula W. Yoon, Sallie B. Freeman, Stephanie L. Sherman, Lisa F. Taft, Yuanhao Gu, Dorothy Pettay, W. Dana Flanders, Muin J. Khoury, and Terry J. Hassold

Letters to the Editor

- 634 Prenatal Diagnosis of 45,X/46,XX**

Lillian Y. F. Hsu

- 635 Reply to Hsu**

Virginia P. Sybert, Dwight Koeberl, and Barbara McGillivray

- 636 Isolated Case of Mental Retardation and Ataxia Due to a De Novo Mitochondrial T8993G Mutation**

I. F. M. de Coö, H. J. M. Smeets, F. J. M. Gabreëls, N. Arts, and B. A. van Oost

638 High Frequency of Mutations in Codon 98 of the Peripheral Myelin Protein Po Gene in 20 French CMT1 Patients

H. Rouger, E. LeGuern, R. Gouider, S. Tardieu, N. Birouk, M. Gugenheim, P. Bouche, Y. Agid, and A. Brice

641 Positive Fragile X Microsatellite Associations Point to a Common Mechanism of Dynamic Mutation Evolution

W. Ted Brown, Nan Zhong, and Carl Dobkin

Book Review**644 The Legacy of Cell Fusion.** Edited by Saimon Gordon

Reviewed by George M. Martin

Announcements**645 Employment and Fellowship Opportunities; Seminar; Conferences; Request for Information; Request for Specimens; Cell Cultures****Errata****648 Segregation and Linkage Analysis of Serum Angiotensin I-Converting Enzyme Levels: Evidence for Two Quantitative-Trait Loci**, by McKenzie et al. (December 1995 [57:1426-1435])**648 Influence of Apolipoprotein E Genotype on the Transmission of Alzheimer Disease in a Community-Based Sample**, by Jarvik et al. (January 1996 [58:191-200])**648 Nutritional Ecogenetics: Homocysteine-Related Arteriosclerotic Vascular Disease, Neural Tube Defects, and Folic Acid**, by Motulsky (January 1996 [58:17-20])**Information for Contributors**

April 1996

1995 ASHG Presidential Address**649 The Challenges and Opportunities of Times of Change**

Judith G. Hall

Original Articles**657 Characterization of the Factor VIII Defect in 147 Patients with Sporadic Hemophilia A: Family Studies Indicate a Mutation Type-Dependent Sex Ratio of Mutation Frequencies**

J. Becker, R. Schwaab, A. Möller-Taube, U. Schwaab, W. Schmidt, H. H. Brackmann, T. Grimm, K. Olek, and J. Oldenburg

671 Glycine Substitutions in the Triple-Helical Region of Type VII Collagen Result in a Spectrum of Dystrophic Epidermolysis Bullosa Phenotypes and Patterns of Inheritance

Angela M. Christiano, John A. McGrath, Kong Chong Tan, and Jouni Uitto

- 682 Compound Heterozygosity for COL7A1 Mutations in Twins with Dystrophic Epidermolysis Bullosa: a Recessive Paternal Deletion/Insertion Mutation and a Dominant Negative Maternal Glycine Substitution Result in a Severe Phenotype**
Angela M. Christiano, Ingrid Anton-Lamprecht, Satoshi Amano, Ulrike Ebschner, Robert E. Burgeson, and Jouni Uitto
- 694 Thiopurine S-Methyltransferase Deficiency: Two Nucleotide Transitions Define the Most Prevalent Mutant Allele Associated with Loss of Catalytic Activity in Caucasians**
Hung-Liang Tai, Eugene Y. Krynetski, Charles R. Yates, Thrina Loennechen, Michael Y. Fessing, Natalia F. Krynetskaia, and William E. Evans
- 703 Genetic and Biochemical Impairment of Mitochondrial Complex I Activity in a Family with Leber Hereditary Optic Neuropathy and Hereditary Spastic Dystonia**
D. D. De Vries, L. N. Went, G. W. Bruyn, H. R. Scholte, R. M. W. Hofstra, P. A. Bolhuis, and B. A. van Oost
- 712 Uroporphyrinogen Decarboxylase: Complete Human Gene Sequence and Molecular Study of Three Families with Hepatoerythropoietic Porphyrria**
M. J. Moran-Jimenez, C. Ged, M. Romana, R. Enriquez de Salamanca, A. Taïeb, G. Topi, L. D'Alessandro, and H. de Verneuil
- 722 Human Homologue Sequences to the *Drosophila dishevelled* Segment-Polarity Gene Are Deleted in the DiGeorge Syndrome**
Antonio Pizzuti, Giuseppe Novelli, Aldo Mari, Antonia Ratti, Alessia Colosimo, Francesca Amati, Donata Penso, Federica Sangiuolo, Giuseppe Calabrese, Giandomenico Palka, Vincenzo Silani, Massimo Gennarelli, Rita Mingarelli, Guglielmo Scarlato, Peter Scambler, and Bruno Dallapiccola
- 730 Non-Mendelian Transmission in Dentatorubral-Pallidoluysian Atrophy and Machado-Joseph Disease: The Mutant Allele Is Preferentially Transmitted in Male Meiosis**
Takeshi Ikeuchi, Shuichi Igarashi, Yoshihisa Takiyama, Osamu Onodera, Mutsuo Oyake, Hiroki Takano, Reiji Koide, Hajime Tanaka, and Shoji Tsuji
- 734 Delineation of a Contiguous Gene Syndrome with Multiple Exostoses, Enlarged Parietal Foramina, Craniofacial Dysostosis, and Mental Retardation, Caused by Deletions on the Short Arm of Chromosome 11**
Oliver Bartsch, Wim Wuyts, Wim Van Hul, Jacqueline T. Hecht, Peter Meinecke, Debra Hogue, Walter Werner, Bernard Zabel, Georg K. Hinkel, Cynthia M. Powell, Lisa G. Shaffer, and Patrick J. Willems
- 743 The Locus for a Novel Syndromic Form of Neuronal Intestinal Pseudoobstruction Maps to Xq28**
Alberto Auricchio, Valeria Brancolini, Giorgio Casari, Peter J. Milla, Virpi V. Smith, Marcella Devoto, and Andrea Ballabio
- 749 Frequent Occurrence of BRCA2 Linkage in Icelandic Breast Cancer Families and Segregation of a Common BRCA2 Haplotype**
Julius Gudmundsson, Gudrun Johannesdottir, Adalgeir Arason, Jon Thor Bergthorsson, Sigurdur Ingvarsson, Valgardur Egilsson, and Rosa Björk Barkardottir
- 757 Linkage of a Gene for Macular Corneal Dystrophy to Chromosome 16**
Jeffery M. Vance, Fridbert Jonasson, Felicia Lennon, Jennifer Sarica, Karim F. Damji, Jennifer Stauffer, Margaret A. Pericak-Vance, and Gordon K. Klintworth
- 763 An Autosomal Locus Predisposing to Multiple Deletions of mtDNA on Chromosome 3p**
Jyrki A. Kaukonen, Patrizia Amati, Anu Suomalainen, Agnes Rötig, Maria-Grazia Piscaglia, Fabrizio Salvi, Jean Weissenbach, Giovanni Fratta, Giacomo Comi, Leena Peltonen, and Massimo Zeviani

- 770 Genetic Mapping of the Hereditary Mixed Polyposis Syndrome to Chromosome 6q**
H. J. W. Thomas, S. C. Whitelaw, S. E. Cottrell, V. A. Murday, I. P. M. Tomlinson, D. Markie, T. Jones, D. T. Bishop, S. V. Hodgson, D. Sheer, J. M. A. Northover, I. C. Talbot, E. Solomon, and W. F. Bodmer
- 777 Familial Cryptic Translocation Resulting in Angelman Syndrome: Implications for Imprinting or Location of the Angelman Gene?**
Leah W. Burke, John E. Wiley, Christopher C. Glenn, Daniel J. Driscoll, Kenneth M. Loud, April J. W. Smith, and Theodore Kushnick
- 785 The Same Molecular Mechanism at the Maternal Meiosis I Produces Mono- and Dicentric 8p Duplications**
Giovanna Floridia, Mauro Piantanida, Antonella Minelli, Claudia Dellavecchia, Clara Bonaglia, Elena Rossi, Giorgio Gimelli, Gianfranco Croci, Fabrizia Franchi, Simone Gilgenkrantz, Paola Grammatico, Leda Dalprá, Stephen Wood, Cesare Danesino, and Orsetta Zuffardi
- 797 Assessment of Aneuploidy for Chromosomes 8, 9, 13, 16, and 21 in Human Sperm by Using Primed In Situ Labeling Technique**
Franck Pellestor, Anne Girardet, Lionel Coignet, Brigitte Andréo, and Jean Paul Charlieu
- 803 Gender Difference in Apolipoprotein E-Associated Risk for Familial Alzheimer Disease: A Possible Clue to the Higher Incidence of Alzheimer Disease in Women**
Haydeh Payami, Sepideh Zareparsy, Kim R. Montee, Gary J. Sexton, Jeffrey A. Kaye, Thomas D. Bird, Chang-En Yu, Ellen M. Wijsman, Leonard L. Heston, Michael Litt, and Gerard D. Schellenberg
- 812 Inherited Susceptibility Determines the Distribution of Dense Low-Density Lipoprotein Subfraction Profiles in Familial Combined Hyperlipidemia**
Sebastian J. H. Bredie, Lambertus A. Kiemeny, Anton F. J. de Haan, Pierre N. M. Demacker, and Anton F. H. Stalenhoef
- 823 Cystic Fibrosis Heterozygote Screening in 5,161 Pregnant Women**
David R. Witt, Catherine Schaefer, Patricia Hallam, Soora Wi, Bruce Blumberg, Andrea Fishbach, Judy Holtzman, Shoshana Kornfeld, Robin Lee, Laurie Nemzer, and Robbin Palmer
- 836 Mapping Quantitative Trait Loci with Extreme Discordant Sib Pairs: Sampling Considerations**
Neil J. Risch and Heping Zhang
- 844 A General Statistical Model for Detecting Complex-Trait Loci by Using Affected Relative Pairs in a Genome Search**
Susan L. Smalley, J. Arthur Woodward, and Christina G. S. Palmer
- 861 Association Studies in Consanguineous Populations**
Emmanuelle Génin and Françoise Clerget-Darpoux
- 867 Nonparametric Simulation-Based Statistics for Detecting Linkage in General Pedigrees**
Sean Davis, Mark Schroeder, Lynn R. Goldin, and Daniel E. Weeks

Letters to the Editor

- 881 Germ-Line *BRCA1* Mutations in Selected Men with Prostate Cancer**
Amelia A. Langston, Janet L. Stanford, Kristine G. Wicklund, Jennifer D. Thompson, Robert G. Blazej, and Elaine A. Ostrander
- 885 The Gene for Nijmegen Breakage Syndrome (V2) Is Not Located on Chromosome 11**
Kenshi Komatsu, Shinya Matsuura, Hiroshi Tauchi, Satoru Endo, Seiji Kodama, Dominique Smeets, Corry Weemaes, and Mitsuo Oshimura

888 Codon 219 Polymorphism of PRNP in Healthy Caucasians and Creutzfeldt-Jakob Disease Patients

Rosella Petraroli and Maurizio Pocchiari

889 CFTR Gene Variant IVS8-5T in Disseminated Bronchiectasis

Pier Franco Pignatti, Cristina Bombieri, Mariagiovanna Benetazzo, Alessandro Casartelli, Elisabetta Trabetti, Lucia Sonia Gilè, Laura Carmen Martinati, Attilio L. Boner, and Maurizio Luisetti

892 Affecteds-Only Linkage Methods Are Not a Panacea

David A. Greenberg, Susan E. Hodge, Veronica J. Vieland, and M. Anne Spence

Obituary

896 Harry Harris (1919-94): In Memoriam

Barton Childs and Richard S. Spielman

Announcements

899 Employment Opportunities; Symposium; Workshop; Cell Cultures; Call for Nominations

Erratum

902 High-Resolution Genetic Mapping of Complex Traits, by L. Kruglyak and E. S. Lander

(May 1995 [56:1212-1223])

Information for Contributors

May 1996

Invited Editorial

903 The FRAXE Syndrome: Is It Time for Routine Screening?

W. Ted Brown

Original Articles

906 A Study of FRAXE in Mentally Retarded Individuals Referred for Fragile X Syndrome (FRAXA) Testing in the United Kingdom

S. J. L. Knight, R. J. Ritchie, L. Chakrabarti, G. Cross, G. R. Taylor, R. F. Mueller, J. Hurst, J. Paterson, J. R. W. Yates, D. J. Dow, and K. E. Davies

914 Guanidinoacetate Methyltransferase Deficiency: The First Inborn Error of Creatine Metabolism in Man

Sylvia Stöckler, Dirk Isbrandt, Folker Hanefeld, Bernhard Schmidt, and Kurt von Figura

923 Differential Effects of FGFR2 Mutations on Syndactyly and Cleft Palate in Apert Syndrome

Sarah F. Slaney, Michael Oldridge, Jane A. Hurst, Gillian M. Morriss-Kay, Christine M. Hall, Michael D. Poole, and Andrew O. M. Wilkie

- 933 Maternally Inherited Cardiomyopathy and Hearing Loss Associated with a Novel Mutation in the Mitochondrial tRNA^{Leu} Gene (G8363A)**
Filippo M. Santorelli, Suk-Chun Mak, Magda El-Schahawi, Carlo Casali, Sara Shanske, Tallie Z. Baram, Ricardo E. Madrid, and Salvatore DiMauro
- 940 The Spectrum of RB1 Germ-Line Mutations in Hereditary Retinoblastoma**
Dietmar R. Lohmann, Birgit Brandt, Wolfgang Höpping, Eberhard Passarge, and Bernhard Horsthemke
- 950 Mucopolysaccharidosis IVA: Four New Exonic Mutations in Patients with N-Acetylgalactosamine-6-Sulfate Sulfatase Deficiency**
Shunji Tomatsu, Seiji Fukuda, Atsushi Yamagishi, Alan Cooper, James E. Wraith, Toshinori Hori, Zenichiro Kato, Naoto Yamada, Kouji Isogai, Kazuko Sukegawa, Naomi Kondo, Yasuyuki Suzuki, Nobuyuki Shimozawa, and Tadao Orii
- 963 Autosomal Recessive Wolfram Syndrome Associated with an 8.5-kb mtDNA Single Deletion**
A. Barrientos, J. Casademont, A. Saiz, F. Cardellach, V. Volpini, A. Solans, E. Tolosa, A. Urbano-Márquez, X. Estivill, and V. Nunes
- 971 Molecular Analysis of Carnitine Palmitoyltransferase II Deficiency with Hepatocardiomyocardial Expression**
Jean-Paul Bonnefont, Franco Taroni, Patrizia Cavadini, Claude Cepanec, Michèle Brivet, Jean-Marie Saudubray, Jean-Paul Leroux, and France Demaugre
- 979 Molecular Characterization of Mitochondrial Trifunctional Protein Deficiency: Formation of the Enzyme Complex Is Important for Stabilization of Both α - and β -Subunits**
Seiichi Ushikubo, Toshifumi Aoyama, Takehiko Kamijo, Ronald J. A. Wanders, Piero Rinaldo, Jerry Vockley, and Takashi Hashimoto
- 989 A *GLRA1* Null Mutation in Recessive Hyperekplexia Challenges the Functional Role of Glycine Receptors**
Wolfram Brune, Ruthild G. Weber, Brigitta Saul, Magnus von Knebel Doeberitz, Caspar Grond-Ginsbach, Klaus Kellermann, Hans-Michael Meinck, and Cord-Michael Becker
- 998 Molecular Analyses of 17p11.2 Deletions in 62 Smith-Magenis Syndrome Patients**
Ramesh C. Juyal, Luis E. Figueroa, Xueya Hauge, Sarah H. Elsea, James R. Lupski, Frank Greenberg, Antonio Baldini, and Pragna I. Patel
- 1008 The Impact of Imprinting: Prader-Willi Syndrome Resulting from Chromosome Translocation, Recombination, and Nondisjunction**
SuEllen Toth-Fejel, Susan Olson, Kristine Gunter, Franklin Quan, Jan Wolford, Bradley W. Popovich, and R. Ellen Magenis
- 1017 An Ancient Common Origin of Aboriginal Australians and New Guinea Highlanders Is Supported by α -Globin Haplotype Analysis**
J. M. Roberts-Thomson, J. J. Martinson, J. T. Norwich, R. M. Harding, J. B. Clegg, and B. Boettcher
- 1025 Mental Status of Females with an FMR1 Gene Full Mutation**
B. B. A. de Vries, A. M. Wieggers, A. P. T. Smits, S. Mohkamsing, H. J. Duivenvoorden, J.-P. Fryns, L. M. G. Curfs, D. J. J. Halley, B. A. Oostra, A. M. W. van den Ouweland, and M. F. Niermeijer

1033 Ascertainment Bias in Estimates of Average Heterozygosity

Alan R. Rogers and Lynn B. Jorde

1042 Genetic Segregation Analyses of Serum IgG2 Levels

Mary L. Marazita, Hong Lu, Margaret E. Cooper, Stephen M. Quinn, Ji-bo Zhang, John A. Burmeister, Joseph V. Califano, Janardan P. Pandey, Harvey A. Schenkein, and John G. Tew

1050 Two-Locus Linkage Analysis of Cutaneous Malignant Melanoma/Dysplastic Nevi

Alisa M. Goldstein, Lynn R. Goldin, Nicholas C. Dracopoli, Wallace H. Clark, Jr., and Margaret A. Tucker

1057 Assessing Familial Aggregation of Age at Onset, by Using Estimating Equations, with Application to Breast Cancer

Li Hsu and Lue Ping Zhao

1072 The Problem of Ascertainment for Linkage Analysis

Veronica J. Vieland and Susan E. Hodge

ASHG/ACMG Report**1085 Diagnostic Testing for Prader-Willi and Angelman Syndromes: Report of the ASHG/ACMG Test and Technology Transfer Committee**

American Society of Human Genetics/American College of Medical Genetics Test and Technology Transfer Committee

Letters to the Editor**1089 A Gene for Premature Ovarian Failure Associated with Eyelid Malformation Maps to Chromosome 3q22-q23**

Patrizia Amati, Paolo Gasparini, Joel Zlotogora, Leopoldo Zelante, Jean Claude Chomel, Alain Kitzis, Josseline Kaplan, and Dominique Bonneau

1092 Limits on Fine Mapping of Complex Traits

Leonid Kruglyak and Eric S. Lander

1093 Likelihood Ratio Tests for Linkage and Linkage Disequilibrium: Asymptotic Distribution and Power

Pak C. Sham, David Curtis, and Charles J. MacLean

1095 Reply to Sham et al.

Joseph D. Terwilliger

Announcements**1097 Fellowship Opportunities; Conferences; Short Course; British Society for Human Genetics; Call for Patients****Erratum****1099 Molecular Mapping of a Recombination Hot Spot Located in the Second Intron of the Human TAP2 Locus, by Cullen et al. (June 1995 [56:1350-1358])****Information for Contributors**

June 1996

Invited Editorials

- 1101 Genetic Control of X Inactivation and Processes Leading to X-Inactivation Skewing**
John W. Belmont
- 1109 Finding Genes on the X Chromosome by Which *Homo* May Have Become *Sapiens***
Gillian Turner

Original Articles

- 1111 Heritability of X Chromosome-Inactivation Phenotype in a Large Family**
Anna K. Naumova, Robert M. Plenge, Lynne M. Bird, Mark Leppert, Kenneth Morgan, Huntington F. Willard, and Carmen Sapienza
- 1120 PPM-X: A New X-Linked Mental Retardation Syndrome with Psychosis, Pyramidal Signs, and Macroorchidism Maps to Xq28**
S. Lindsay, M. Splitt, S. Edney, T. P. Berney, S. J. L. Knight, K. E. Davies, O. O'Brien, M. Gale, and J. Burn
- 1127 Identification, Expression, and Biochemical Characterization of N-Acetylgalactosamine-4-Sulfatase Mutations and Relationship with Clinical Phenotype in MPS-VI Patients**
Tom Litjens, Doug A. Brooks, Christoph Peters, Gary J. Gibson, and John J. Hopwood
- 1135 Mutational and Protein Analysis of Patients and Heterozygous Women with X-Linked Adrenoleukodystrophy**
Valérie Feigenbaum, G     Lombard-Platet, Sylvie Guidoux, Claude-Olivier Sarde, Jean-Louis Mandel, and Patrick Aubourg
- 1145 Mutation in and Lack of Expression of Tyrosinase-Related Protein-1 (TRP-1) in Melanocytes from an Individual with Brown Oculocutaneous Albinism: A New Subtype of Albinism Classified as "OCA3"**
Raymond E. Boissy, Huiquan Zhao, William S. Oetting, Lisa M. Austin, Scott C. Wildenberg, Ying L. Boissy, Yang Zhao, Richard A. Sturm, Vincent J. Hearing, Richard A. King, and James J. Nordlund
- 1157 A Mutation Causing Alport Syndrome with Tardive Hearing Loss Is Common in the Western United States**
David F. Barker, Charles J. Pruchno, Xia Jiang, Curtis L. Atkin, Edwin M. Stone, Joyce C. Denison, Pamela R. Fain, and Martin C. Gregory
- 1166 Two Distinct Origins of a Common *BRCA1* Mutation in Breast-Ovarian Cancer Families: A Genetic Study of 15 185delAG-Mutation Kindreds**
David B. Berman, Josephine Wagner-Costalas, David C. Schultz, Henry T. Lynch, Mary Daly, and Andrew K. Godwin
- 1177 Substitution of a Conserved Cysteine-996 in a Cysteine-Rich Motif of the Laminin α 2-Chain in Congenital Muscular Dystrophy with Partial Deficiency of the Protein**
Marja Nissinen, Anne Helbling-Leclerc, Xu Zhang, Teresinha Evangelista, Haluk Topaloglu, Corinne Cruaud, Jean Weissenbach, Michel Fardeau, Fernando M. S. Tom    , Ketty Schwartz, Karl Tryggvason, and Pascale Guicheney

- 1185 A Novel Mutation in the Putative DNA Helicase *XH2* Is Responsible for Male-to-Female Sex Reversal Associated with an Atypical Form of the ATR-X Syndrome**
A. Ion, L. Telvi, J. L. Chaussain, F. Galacteros, J. Valayer, M. Fellous, and K. McElreavey
- 1192 X-Linked Alport Syndrome: An SSCP-Based Mutation Survey over All 51 Exons of the *COL4A5* Gene**
Alessandra Renieri, Mirella Bruttini, Lucia Galli, Paola Zanelli, Tauro Neri, Sandro Rossetti, Alberto Turco, Nina Heiskari, Jing Zhou, Rosanna Gusmano, Laura Massella, Giovanni Banfi, Francesco Scolari, Adalberto Sessa, Gianfranco Rizzoni, Karl Tryggvason, Pier Franco Pignatti, Mario Savi, Andrea Ballabio, and Mario De Marchi
- 1205 Mutations and Phenotype in Isolated Glycerol Kinase Deficiency**
Ann P. Walker, Françoise Muscatelli, Amanda N. Stafford, Jamel Chelly, Niklas Dahl, Hans K. Blomquist, Joris Delanghe, Patrick J. Willems, Beat Steinmann, and Anthony P. Monaco
- 1212 Somatic Mosaicism of Expanded CAG Repeats in Brains of Patients with Dentatorubral-Pallidoluysian Atrophy: Cellular Population-Dependent Dynamics of Mitotic Instability**
Hiroki Takano, Osamu Onodera, Hitoshi Takahashi, Shuichi Igarashi, Mitsunori Yamada, Mutsuo Oyake, Takeshi Ikeuchi, Reiji Koide, Hajime Tanaka, Kiyoshi Iwabuchi, and Shoji Tsuji
- 1223 Recombination Hot Spot in a 3.2-kb Region of the Charcot-Marie-Tooth Type 1A Repeat Sequences: New Tools for Molecular Diagnosis of Hereditary Neuropathy with Liability to Pressure Palsies and of Charcot-Marie-Tooth Type 1A**
Judith Lopes, Eric LeGuern, Riadh Gouider, Sandrine Tardieu, Nacer Abbas, Nazha Birouk, Michel Gugenheim, Pierre Bouche, Yves Agid, Alexis Brice, and the French CMT Collaborative Research Group
- 1231 Molecular Analysis of Recombination in a Family with Duchenne Muscular Dystrophy and a Large Pericentric X Chromosome Inversion**
Vandana Shashi, Wendy L. Golden, Patricia S. Allinson, Susan Halloran Blanton, Christopher von Kap-Herr, and Thaddeus E. Kelly
- 1239 Testing the Feasibility of DNA Typing for Human Identification by PCR and an Oligonucleotide Ligation Assay**
Claire Delahunty, Wendy Ankener, Qiang Deng, Jimmy Eng, and Deborah A. Nickerson
- 1247 Progressive Myoclonus Epilepsy *EPM1* Locus Maps to a 175-kb Interval in Distal 21q**
Kimmo Virtaneva, Jinmin Miao, Ann-Liz Träskelin, Nancy Stone, Janet A. Warrington, Jean Weissenbach, Richard M. Myers, David R. Cox, Pertti Sistonen, Albert de la Chapelle, and Anna-Elina Lehesjoki
- 1254 Linkage of Congenital Recessive Deafness (Gene *DFNB10*) to Chromosome 21q22.3**
Batsheva Bonné-Tamir, Anita L. DeStefano, Christine E. Briggs, Ronald Adair, Barbara Franklyn, Sari Weiss, Michael Korostishevsky, Moshe Frydman, Clinton T. Baldwin, and Lindsay A. Farrer
- 1260 Deletion Mapping of Gliomas Suggests the Presence of Two Small Regions for Candidate Tumor-Suppressor Genes in a 17-cM Interval on Chromosome 10q**
Ruth Albarosa, Bruno M. Colombo, Luca Roz, Ivana Magnani, Bianca Pollo, Nicola Cirenei, Cristiana Giani, Anna Maria Fuhrman Conti, Stefano DiDonato, and Gaetano Finocchiaro
- 1268 Identification of New Polymorphisms of the Angiotensin I-Converting Enzyme (*ACE*) Gene, and Study of Their Relationship to Plasma *ACE* Levels by Two-QTL Segregation-Linkage Analysis**
Eric Villard, Laurence Tiret, Sophie Visvikis, Roger Rakotovo, François Cambien, and Florent Soubrier
- 1279 Affected-Sib-Pair Analyses Reveal Support of Prior Evidence for a Susceptibility Locus for Bipolar Disorder, on 21q**
Sevilla D. Detera-Wadleigh, Judith A. Badner, Lynn R. Goldin, Wade H. Berrettini, Alan R. Sanders, Denise Y. Rollins, Gordon Turner, Tracy Moses, Hafez Haerian, David Muniec, John I. Nurnberger, Jr., and Elliot S. Gershon

- 1286 Toward Localization of the Werner Syndrome Gene by Linkage Disequilibrium and Ancestral Haplotyping: Lessons Learned from Analysis of 35 Chromosome 8p11.1-21.1 Markers**
Katrina A. B. Goddard, Chang-En Yu, Junko Oshima, Tetsuro Miki, Jun Nakura, Charles Piusan, George M. Martin, Gerard D. Schellenberg, Ellen M. Wijsman, and members of the International Werner's Syndrome Collaborative Group
- 1303 β -Globin Haplotype Analysis Suggests That a Major Source of Malagasy Ancestry Is Derived from Bantu-Speaking Negroids**
Rachel Hewitt, Amanda Krause, Andrea Goldman, Gwyn Campbell, and Trefor Jenkins
- 1309 The Genetic Relationship between the Finns and the Finnish Saami (Lapps): Analysis of Nuclear DNA and mtDNA**
Päivi Lahermo, Antti Sajantila, Pertti Sistonen, Matti Lukka, Pertti Aula, Leena Peltonen, and Marja-Liisa Savontaus
- 1323 Descent Graphs in Pedigree Analysis: Applications to Haplotyping, Location Scores, and Marker-Sharing Statistics**
Eric Sobel and Kenneth Lange
- 1338 Conclusions of LOD-Score Analysis for Family Data Generated under Two-Locus Models**
Marie-Hélène Dizier, Marie-Claude Babron, and Françoise Clerget-Darpoux
- 1347 Parametric and Nonparametric Linkage Analysis: A Unified Multipoint Approach**
Leonid Kruglyak, Mark J. Daly, Mary Pat Reeve-Daly, and Eric S. Lander
- 1364 High Male:Female Ratio of Germ-Line Mutations: An Alternative Explanation for Postulated Gestational Lethality in Males in X-Linked Dominant Disorders**
George H. Thomas

Letters to the Editor

- 1369 North and South Amerindians May Have the Same Major Founder Y Chromosome Haplotype**
Fabrício R. Santos, Luís Rodríguez-Delfin, Sérgio D. J. Pena, John Moore, and Kenneth M. Weiss
- 1370 The Gene for Replication Factor C Subunit 2 (RFC2) Is within the 7q11.23 Williams Syndrome Deletion**
Risa Peoples, Luis Perez-Jurado, Yu-Ker Wang, Paige Kaplan, and Uta Francke
- 1373 Maple Syrup Urine Disease: The E1 β Gene of Human Branched-Chain α -Ketoacid Dehydrogenase Complex Has 11 Rather than 10 Exons, and the 3' UTR in One of the Two E1 β mRNAs Arises from Intronic Sequences**
Jacinta L. Chuang, Rody P. Cox, and David T. Chuang
- 1377 Deletion Mapping of 22q11 in CATCH22 Syndrome: Identification of a Second Critical Region**
Hiroki Kurahashi, Takahiro Nakayama, Yuko Osugi, Etsuko Tsuda, Mitsuo Masuno, Kiyoshi Imaizumi, Tetsuro Kamiya, Tetsuya Sano, Shintaro Okada, and Isamu Nishisho
- 1381 Detection of Linkage to Affective Disorders in the Catalogued Amish Pedigrees: A Reply to Pauls et al.**
Elliot S. Gershon, Lynn R. Goldin, Judith A. Badner, and Wade H. Berrettini
- 1384 Chromosome 18 Markers: Linked or Not Linked to Bipolar Affective Disorders in the Old Order Amish? A Reply to Gershon et al.**
David L. Pauls, Jurg Ott, Steven M. Paul, Cleona R. Allen, Cathy S. J. Fann, John P. Carulli, Kathleen M. Falls, Christine A. Bouthillier, Thomas C. Gravius, Tim P. Keith, Janice A. Egeland, and Edward I. Ginns

Announcements

- 1386 Employment and Fellowship Opportunities; Seminar; Call for Nominations; Request for Proposals; Cell Cultures; Call for Patients**

Erratum

- 1389 Relative Risk of Alzheimer Disease and Age-at-Onset Distributions, Based on *APOE* Genotypes among Elderly African Americans, Caucasians, and Hispanics in New York City, by Tang et al. (March 1996 [58:574–584])**

- 1390 Author Index for Volume 58**

- 1397 Subject Index for Volume 58**

- 1405 Contents of Volume 58**

Information for Contributors